



## AGL gene

amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase

### Normal Function

The *AGL* gene provides instructions for making the glycogen debranching enzyme. This enzyme is involved in the breakdown of a complex sugar called glycogen, which is a major source of stored energy in the body. Glycogen is made up of several molecules of a simple sugar called glucose. Some glucose molecules are linked together in a straight line, while others branch off and form side chains. The glycogen debranching enzyme is involved in the breakdown of these side chains. The branched structure of glycogen makes it more compact for storage and allows it to break down more easily when it is needed for fuel.

The *AGL* gene provides instructions for making several different versions (isoforms) of the glycogen debranching enzyme. These isoforms vary by size and are active (expressed) in different tissues.

### Health Conditions Related to Genetic Changes

#### glycogen storage disease type III

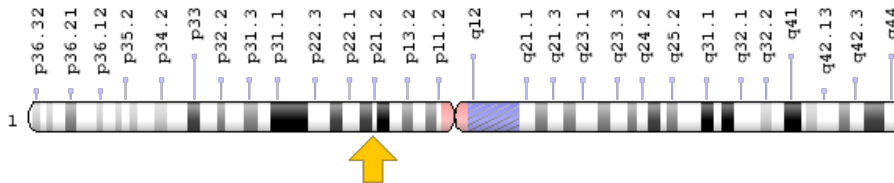
Approximately 100 mutations in the *AGL* gene have been found to cause glycogen storage disease type III (also called GSDIII or Cori disease). Most of these mutations lead to a premature stop signal in the instructions for making the glycogen debranching enzyme, resulting in a nonfunctional enzyme. As a result, the side chains of glycogen molecules cannot be removed and abnormal, partially broken down glycogen molecules are stored within cells. A buildup of abnormal glycogen damages organs and tissues throughout the body, particularly the liver and muscles, leading to the signs and symptoms of GSDIII.

Mutations in the *AGL* gene can affect different isoforms of the enzyme, depending on where the mutations are located in the gene. For example, mutations that occur in a part of the *AGL* gene called exon 3 affect the isoform that is primarily expressed in the liver. These mutations almost always lead to GSD type IIIb, which is characterized by liver problems.

## Chromosomal Location

Cytogenetic Location: 1p21.2, which is the short (p) arm of chromosome 1 at position 21.2

Molecular Location: base pairs 99,850,077 to 99,924,023 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- amylo-1, 6-glucosidase, 4-alpha-glucanotransferase
- GDE
- GDE\_HUMAN
- glycogen debrancher
- glycogen debranching enzyme

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): A Debranching Enzyme Also Is Needed for the Breakdown of Glycogen  
<https://www.ncbi.nlm.nih.gov/books/NBK22467/#A2919>
- Washington University, St. Louis: Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/msys/glycogen.html#deb>

### GeneReviews

- Glycogen Storage Disease Type III  
<https://www.ncbi.nlm.nih.gov/books/NBK26372>

### Genetic Testing Registry

- GTR: Genetic tests for AGL  
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=178%5Bgeneid%5D>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28AGL%5BTIAB%5D%29+OR+%28glycogen+debranching+enzyme%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>

### OMIM

- AMYLO-1,6-GLUCOSIDASE, 4-ALPHA-GLUCANOTRANSFERASE  
<http://omim.org/entry/610860>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_AGL.html](http://atlasgeneticsoncology.org/Genes/GC_AGL.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=AGL%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=321](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=321)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/178>
- UniProt  
<http://www.uniprot.org/uniprot/P35573>

### **Sources for This Summary**

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